

Prenatal Detection of Associated Extra Cardiac Malformations with Congenital Heart Defects on Obstetric Ultrasound

Pankaj Kaler¹, P S Mishrikotkar², Asmita Suryawanshi³

¹Resident, Department of Radiology, MGM Medical College Aurangabad (MS) India.

²Professor and HOD, Department of Radiology, MGM Medical College Aurangabad (MS) India.

³Associate Professor, Department of Radiology, MGM Medical College Aurangabad (MS) India.

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ABSTRACT

Background: The association of CHD (congenital heart defects) with major extra-cardiac malformations (ECMs) is common, and extensive screening for associated anomalies is recommended when a CHD is identified in a fetus. **Aims & Objectives:** To study the association between congenital heart defects and extra cardiac malformations on obstetric ultrasound. To detect various patterns of extra cardiac malformations. To detect any suspected syndromic anomaly case.

Methods: The study was conducted from October 2017 to October 2019 (24 months).The population for this study included all pregnant females who are referred from obstetrics and gynaecology department and thus sent to department of radio diagnosis for ante natal sonographic examination. **Results:** Out of 31 diagnosed cases of congenital heart defects (CHD), 9 cases (29.03%) were having associated extra cardiac malformations (ECM). Multiple extra cardiac malformations were seen in two cases (22.2%), rest of the cases were having only single extra cardiac malformation (77.8%). Associated extra cardiac malformations were craniofacial anomalies in 4 cases (44.4%), gastro intestinal in 2 cases (22.2%), renal anomalies in 2 cases (22.2%),central nervous system in 1 case (11.1%), respiratory system anomaly in 1 case (11.1%) and lympho vascular anomaly in 1 case (11.1%). **Conclusions:** The association of congenital heart defects with major extra cardiac malformations and suspected genetic syndromes is well documented. So whenever any malformation is detected on obstetric ultrasound prenatally, a detail study should be done to look for any other associated anomaly.

Keywords: Congenital heart defects (CHD), extra cardiac malformations (ECM), fetal heart defect.

1

INTRODUCTION

The proportion of congenital heart diseases (CHDs) associated with extra-cardiac malformations (ECMs) and cytogenetic anomalies has been extensively reported.^[1-5] The association of CHD with major extra-cardiac malformations (ECMS) is common, and extensive screening for associated anomalies is recommended when a CHD is identified in a fetus.^[6,7] Intuitively, the association of a CHD with another anomaly makes prenatal counselling more complex, and might influence parental decision regarding continuation of pregnancy.^[8,9]

The prevalence of CHD has been increasing due to the greater detection of Minor defects by the Doppler echocardiography, which has been widely used. In addition. Extra-cardiac malformations (EM), and/or associated with genetic syndromes, are observed from 7 to 50% of the patients with CHD, and impose

a greater risk of morbidity and mortality to these patients, in addition to increasing the risks of surgical correction. Also, such patients may need surgical procedures or intensive care regardless of the heart condition.^[10-14]

MATERIALS & METHODS

The was a prospective study conducted from October 2017 to October 2019 (24 months) in the deptment of radiology at MGM Medical College and Hospital, Aurangabad. All pregnant females who are referred from obstetrics and gynaecology department and thus sent to department of Radio diagnosis for antenatal sonographic examination. A complete antenatal ultrasound examination of pregnant women included in the study will be done using gray scale & colour duplex examination. Philips I/U 22 and Volusion (E8) ultrasound machines were used for the purpose of scanning.

Eligibility Criteria:-

Inclusion criteria:

- All pregnant women coming for antenatal sonographic examination during the second trimester

Name & Address of Corresponding Author

Dr Pankaj Kaler
Resident,
Department Of Radiology,
MGM Medical College
Aurangabad (MS) India.

Kaler et al; Prenatal Detection of Associated Extra Cardiac Malformations with Congenital Heart Defects

at Department of Radio diagnosis, MGM Aurangabad.

- Informed written consent was obtained.

Exclusion criteria:

- Obese patients
- Patients who are not willing to give consent.
- Improper fetal position while examination.

RESULTS

Out of 31 diagnosed cases of fetal heart defects, 9 cases (29.03%) were having associated extra cardiac malformations. Multiple extra cardiac malformations were seen in two cases (22.2%), rest of the cases were having only single extra cardiac malformation (77.8%). Associated extra cardiac malformations were craniofacial in 4 cases (44.4%), gastro intestinal in 2 cases(22.2%), renal anomalies in 2 cases (22.2%),central nervous system in 1 case (11.1%), respiratory system anomaly in 1 case (11.1%) and lympho vascular anomaly in 1 case (11.1%). Most common congenital heart defects having extra cardiac malformations were Ventricular septa defects in 4 cases (44.4%) and Hypoplastic left heart syndromes (HLHS) in 4 cases (44.4%). Rest were tetralogy of fallot (TOF) in 1 case (11.1%) and persistent superior vena cava with atrial septal defect in one case (11.1%) Suspected syndromic anomalies were seen in 4 cases (44.4 %) of fetal heart defects and rest of the 5 cases(55.6%) were non syndromic anomalies.

Table 1: Proportion Of Extracardiac Malformation

Total no of CHD	Total no of ECM	Proportion
31	9	29.03 %

Total 31 patients were diagnosed with fetal heart defects, out of them 9 cases (29.03%) were having associated extra cardiac malformations.

Table 2: Patterns Of Extra Cardiac Malformations

System	type	No. Of cases	Percentage
Craniofacial	Absent Nasal Bone	2	44.4 %
	Hypoplastic Nasal Bone	1	
	Cleft Lip And Cleft Palate	1	
Gastrointestinal	Omphalocele	1	22.2 %
	Diaphragmatic Hernia	1	
Central Nervous System	Encephalocole	1	11.1 %
Respiratory	Hypoplastic Lungs	1	11.1%
Renal	Left Hydronephrosis ?Puj Obstruction	1	22.2%
	Right Hydronephrosis	1	
Lymphovascular	Cystic Hygroma	1	11.1%
Musculoskeletal	Biltaral Club Foot	1	11.1 %

Associated extra cardiac malformations were craniofacial anomalies in 4 cases (44.4%), gastro intestinal anomalies in 2 cases(22.2%), renal anomalies in 2 cases (22.2%),central nervous system in 1 case (11.1%), respiratory system anomaly in 1 case (11.1%) and lympho vascular anomaly in 1 case (11.1%). 2 diagnosed cases of fetal heart defects were having more than one extra cardiac malformations.

Table 3: Types Of Chd And Associated Ecm

Type Of CHD	Type of ECM
Hypoplastic left heart syndrome	Hypoplastic nasal bone
Hypoplastic left heart syndrome	Diaphragmatic hernia
Hypoplastic left heart syndrome	Encephalocole
Hlhs,vsd,tricuspid regurgitation	Hypoplastic lungs
Tetralogy of fallot	Omphalocele Bilateral club foot Cleft lip and cleft palate
Ventricular septal defect (vsd)	Hypoplastic nasal bone
Vsd and common truncus arteriosus (cta)	Left hydronephrosis ?puj obstruction
Vsd	Cystic hygroma & hydrops fetalis & right hydronephrosis
Persistent svc & asd	Absent nasal bone

Most common fetal heart defects associated with extra cardiac malformations were ventricular septal defects in 4 cases (44.4%) and Hypoplastic left heart syndrome in 4 cases (44.4%). Rest were tetralogy of fallot (TOF) in 1 case (11.1%) and persistent superior vena cava with atrial septal defect in one case (11.1%)

Table 4: Single/Multiple Extracardiac Malformation

ECM	No. Of cases	Percentage
Single	7	77.8 %
Multiple	2	22.2 %

Multiple extra cardiac malformations were seen in two cases (22.2%), rest of the cases were having only single extra cardiac malformation (77.8%).

Table 5: Suspected Syndromic/Nonsyndromic Anomalies

	Fetaures	No. Of cases	Percentage
Suspected Syndrome	Absent/Hypoplastic Nasal Bone (Down's) Cystic Hygroma With Right Hydronephrosis And Hydrops Fetalis (Noonan's)	3 1	44.4 %
Non Syndromic		5	55.6 %

Suspected syndromic anomalies were seen in 4 cases (44.4 %) of fetal heart defects and rest of the 5 cases (55.6%) were non syndromic anomalies.



Figure 1: Omphalocole:- Anterior Abdominal Wall Defect With Herniation Of Omentum.



Figure 2: Club Foot



Figure 3: Diaphragmatic hernia



Figure 4: Encephalocole



Figure 5: Absent Nasal Bone

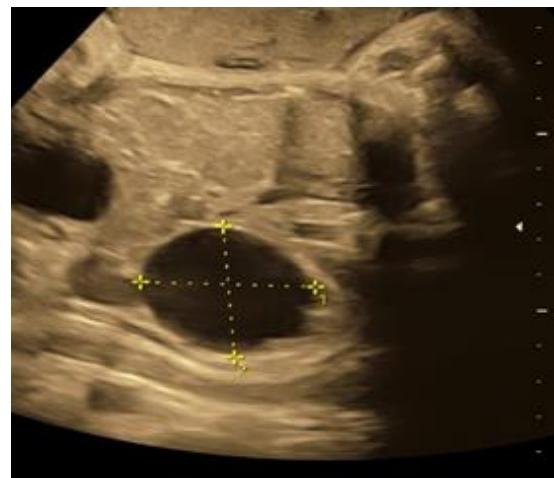


Figure 6: Hydronephrosis ?Puj Obstruction



Figure 7: Hypoplastic lungs with pleural effusion and ascitis.

DISCUSSION

In our study Total 31 patients were diagnosed with fetal heart defects, out of them 9 cases (29.03%) were having associated extra cardiac malformations. These findings are comparable with study done by Ferencz et al^[15] in 1987 where association of extracardiac malformation with congenital heart defects cases was present in 26.8 % cases and with

Kaler et al; Prenatal Detection of Associated Extra Cardiac Malformations with Congenital Heart Defects

study done by Eskedal et al in 2004 where association was present in 20% cases.^[16]

Associated extra cardiac malformations were craniofacial anomalies in 4 cases (44.4%), gastro intestinal anomalies in 2 cases (22.2%), renal anomalies in 2 cases (22.2%), central nervous system in 1 case (11.1%), respiratory system anomaly in 1 case (11.1%) and lympho vascular anomaly in 1 case (11.1%). These findings are comparable with study done by Gucer et al in which craniofacial anomalies were the most common anomalies associated with cardiac malformation cases.^[17]

Study done by Eskedal et al in 2004 gastrointestinal anomalies were the most common associated extracardiac malformations. In our study gastrointestinal anomalies were the second most common associated anomalies.^[16]

Most common fetal heart defects associated with extra cardiac malformations were ventricular septal defects in 4 cases (44.4%) and Hypoplastic left heart syndrome in 4 cases (44.4%). These findings are comparable with the study done by Calzolari et al in 2003 in which the most common cardiac defects those were associated with extracardiac malformations were ventricular septal defects, complex cardiac defects and ASD.^[18]

CONCLUSION

The association of congenital heart defects with major extra cardiac malformations and suspected genetic syndromes is well documented. So whenever any malformation is detected on obstetric ultrasound prenatally, a detail study should be done to look for any other associated anomaly. The prognosis of the fetus with multiple malformations is always poor so detection of the anomalies at appropriate time is very important in decision making for termination of pregnancy.

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